CONDITION

Muscular dystrophy

Muscular dystrophy is a group of inherited conditions that gradually weaken muscles and may affect the heart and breathing.

WHAT IT IS
Inherited conditions causing progressive muscle weakness

BODY SYSTEM
Muscles, heart, lungs, and nervous system

MAIN SYMPTOMS
Muscle weakness, frequent falls, difficulty climbing stairs, muscle stiffness

WHAT CAUSES IT
Inherited changes in genes needed for healthy muscles

HOW IT IS DIAGNOSED
Genetic testing, sometimes muscle biopsy

MAIN TREATMENT
Rehabilitation, medicines, mobility aids, heart and breathing support

CAN IT BE CURED?
No - treatments manage effects and may slow progression

IS IT AN EMERGENCY?
Sometimes - severe breathing difficulty needs emergency care

LONG-TERM OUTLOOK
Progressive weakness; pace and complications differ by type

Overview

Muscular dystrophy is the name for a group of inherited conditions that cause muscles to become weaker over time. Genetic changes prevent muscle cells from making or using certain proteins correctly. This makes the cells more easily damaged and less able to repair themselves.

There are many types. They differ in the muscles they affect, the age symptoms begin, and how quickly weakness progresses. Some mainly affect the hips and shoulders. Others affect the face, hands, lower legs, eyes, swallowing muscles, heart, or breathing muscles.

Common types include Duchenne, Becker, myotonic, facioscapulohumeral, limb-girdle, congenital, Emery-Dreifuss, and oculopharyngeal muscular dystrophy. Each has its own pattern and genetic cause.

Some types begin in infancy or childhood, while others do not become noticeable until adulthood. Certain forms progress slowly and allow long-term independence. Others cause major mobility, heart, or breathing problems. An exact diagnosis matters because monitoring and treatment are tailored to the type.

Symptoms

Progressive muscle weakness is the main feature. "Progressive" means that it tends to increase over time, although the pace is different for each type and person.

Early signs in a child may include:

In teenagers and adults, early changes may include difficulty lifting objects, raising the arms, climbing stairs, or walking long distances. Weak shoulder muscles can cause the shoulder blades to stick out. Weak facial muscles may make it hard to close the eyes fully, whistle, or use facial expressions.

Other symptoms depend on the type. Myotonic dystrophy can cause myotonia , meaning muscles do not relax promptly after contracting. For example, it may be difficult to release a hand grip. Oculopharyngeal muscular dystrophy commonly affects the eyelids and swallowing.

Some people develop muscle shortening, called contractures , which limits joint movement. Muscle cramps, fatigue, balance problems, and pain can occur, although pain is not always a main symptom.

Muscular dystrophy may also affect areas beyond the muscles used for movement. Possible effects include:

Not everyone develops all of these symptoms.

- Delayed sitting, standing, walking, or running
- Frequent falls or difficulty getting up from the floor
- Walking on the toes or with a waddling movement
- Difficulty climbing stairs or keeping up with other children
- Enlarged-looking calf muscles caused partly by fat and scar tissue
- Weak breathing muscles or an ineffective cough
- Abnormal heart rhythm or a weakened heart muscle
- Difficulty chewing, swallowing, or speaking
- Curvature of the spine

- Learning, attention, or emotional difficulties in some types

Causes and risk factors

Muscular dystrophy is caused by changes, also called variants or mutations, in genes that help muscles develop, function, or repair themselves. Different genetic changes cause different types.

The changed gene may be inherited from one or both parents. The inheritance pattern can be:

Sometimes a genetic change starts for the first time in an egg, sperm, or early embryo. This is called a new or de novo variant. A person can therefore have muscular dystrophy without a known family history.

Family history is the main risk factor, but its absence does not rule out the condition. Muscular dystrophy is not caused by exercise, diet, parenting, infection, or anything someone did during pregnancy.

- X-linked: The changed gene is on the X chromosome. Duchenne and Becker muscular dystrophy usually affect boys and men more severely, although girls and women who carry the change can have symptoms.
- Autosomal dominant: One changed copy of a gene can cause the condition.
- Autosomal recessive: Both copies of a gene must be changed, usually with one inherited from each parent.

Diagnosis

A clinician will ask about symptoms, childhood development, walking, falls, and family history. The examination looks at muscle strength, movement, reflexes, posture, and the pattern of weakness.

Blood tests often measure creatine kinase , or CK. This enzyme leaks into the blood when muscle cells are damaged. A high CK level can support suspicion of a muscle condition, but it does not identify the exact type by itself.

Genetic testing is central to diagnosis. It looks for gene changes known to cause muscular dystrophy. Finding a relevant variant can confirm the type, guide family testing, and show whether certain targeted treatments might be suitable. Sometimes testing does not find a clear answer immediately, and broader genetic analysis may be considered.

A muscle biopsy may be needed when genetic results are uncertain. A small sample of muscle

is examined for changes in muscle fibres and proteins. Electromyography, which measures electrical activity in muscles, is sometimes used to help distinguish muscle weakness from a nerve problem.

Other tests assess possible complications rather than confirming the condition. These may include:

Genetic counselling can help you understand the result, what it may mean for relatives, and the chance of passing the condition to a child.

- An electrocardiogram and heart imaging
- Breathing tests and sleep studies
- Swallowing assessments
- Bone-strength or spine imaging

Treatment and management

There is currently no treatment that cures all forms of muscular dystrophy. Care aims to maintain movement and independence, slow some effects where possible, and protect the heart, lungs, bones, and joints.

A care team may include neurology, rehabilitation, physiotherapy, heart, breathing, bone health, genetics, nutrition, and mental health specialists.

Physiotherapy can help preserve joint movement and manage contractures. A therapist may recommend stretching and suitable physical activity. Moderate activity may support general health, but exhausting or high-impact exercise can place extra stress on fragile muscles. Advice should be matched to the type and level of weakness.

Occupational therapy focuses on daily activities such as dressing, bathing, school, work, and using technology. Braces, wheelchairs, scooters, standing equipment, or home adaptations can improve safety and independence. Using a mobility aid is not a treatment failure; it may conserve energy and support participation.

Corticosteroid medicines can help preserve strength and function for a period in many people with Duchenne muscular dystrophy. They can also cause important side effects, so their benefits and monitoring need to be discussed with the specialist team.

Gene-targeted or RNA-targeted treatments are available in some places for selected people with Duchenne muscular dystrophy. Eligibility may be based on the exact genetic variant, age, stage of the condition, and local approval. These treatments are not cures, and their likely benefits and uncertainties differ.

Management may also include:

Before surgery or a general anaesthetic, tell the surgical and anaesthetic teams about the muscular dystrophy. Muscle, heart, and breathing problems can affect anaesthetic planning.

- Medicines and monitoring for heart problems
- Assisted coughing or non-invasive breathing support
- Nutrition and swallowing support
- Treatment for weak bones or fractures
- Surgery for severe contractures or spinal curvature
- Educational, psychological, and social support

Complications

Weakness can lead to falls, reduced mobility, contractures, and loss of independence with some activities. Spinal curvature may affect sitting balance or breathing. Limited movement and some medicines can contribute to weak bones and fractures.

A weakened heart muscle, called cardiomyopathy , can make it harder for the heart to pump. Some types also cause abnormal heart rhythms. These problems may develop before obvious symptoms, which is why planned heart monitoring matters.

Weak breathing muscles can cause shallow breathing, especially during sleep. Signs may include morning headaches, disturbed sleep, daytime sleepiness, difficulty concentrating, or repeated chest infections. A weak cough can make it harder to clear mucus.

Swallowing weakness may cause coughing during meals, food sticking, weight loss, dehydration, or food entering the airway. This last problem is called aspiration and can lead to a chest infection.

Fatigue, changes in appearance or independence, and uncertainty about progression can affect emotional well-being. Support may be helpful for both the person with muscular dystrophy and family members.

Prevention

Inherited muscular dystrophy cannot be prevented through lifestyle changes. Exercise, supplements, or diet cannot remove the genetic change that causes it.

If a genetic cause has been identified in a family, genetic counselling can explain reproductive

options. These may include testing during pregnancy, testing embryos created through in vitro fertilisation, donor eggs or sperm, or choosing not to test. These are personal options rather than requirements.

Although the condition itself cannot usually be prevented, planned monitoring and treatment may reduce some complications. This includes attention to heart and breathing health, safe mobility, joint movement, bone strength, swallowing, and nutrition.

Living with it and what to expect

The course over months and years is strongly shaped by the type of muscular dystrophy. Duchenne usually begins in early childhood and causes increasing weakness. Becker often progresses more slowly. Some limb-girdle and facioscapulohumeral forms remain mild for many years, while others cause substantial mobility problems. Adult-onset types may also affect the heart, breathing, swallowing, or other body systems.

Abilities can change gradually rather than from day to day. Tasks such as climbing stairs, lifting the arms, walking outdoors, or transferring from a chair may require more effort or new equipment. Rehabilitation and mobility aids can help you adapt as needs change.

Regular appointments may continue even when symptoms feel stable. Heart or breathing changes can develop without being obvious at first. The recommended checks and their timing are based on the exact type, age, symptoms, and treatments.

Many people with muscular dystrophy attend school, work, form relationships, raise families, travel, and take part in their communities. Practical support may include accessible education or employment, personal assistance, transport help, and financial or disability services.

Life expectancy differs widely. Some types have little effect on lifespan. Others can shorten life because of heart or breathing complications. Modern monitoring, assisted ventilation, and heart treatment have improved outcomes for several forms, but no individual course can be promised.

When to get medical help

Arrange a routine medical assessment if you or your child has gradually increasing muscle weakness, frequent unexplained falls, delayed movement milestones, difficulty climbing stairs, or a family history of muscular dystrophy. These symptoms can have many causes and need proper assessment.

Contact the usual clinical team promptly for:

Seek emergency help for severe difficulty breathing, blue or grey lips, inability to stay awake, sudden chest pain, fainting, or choking that blocks breathing. Sudden severe weakness can also require emergency assessment.

- New or worsening breathlessness, especially when lying down
- Morning headaches, daytime sleepiness, or a noticeably weaker cough
- Repeated choking, coughing during meals, or unexplained weight loss
- New palpitations, swelling, or a clear decline in mobility
- Fever and chest symptoms when coughing is weak

Common questions

Is muscular dystrophy fatal?

Some types can shorten life, particularly when they seriously affect the heart or breathing muscles. Other types progress slowly and may have little effect on lifespan. The exact diagnosis and access to monitoring and supportive treatment are important.

Can women and girls have muscular dystrophy?

Yes. Muscular dystrophy can affect people of any sex. X-linked forms such as Duchenne and Becker are usually more severe in boys and men, but girls and women who carry the genetic change can develop muscle weakness, heart problems, or other symptoms.

Can muscular dystrophy skip a generation?

It can appear to. Someone may carry a genetic change without obvious symptoms and pass it on. Recessive forms can also appear unexpectedly when both parents carry the same changed gene. Genetic testing and counselling can clarify the pattern in a family.

Is muscular dystrophy painful?

The muscle weakness itself is not always painful. Some people have cramps, joint discomfort, back pain, or pain caused by posture, contractures, falls, or reduced movement. New or persistent pain should be assessed rather than assumed to be part of the condition.

Is exercise good for muscular dystrophy?

Appropriate activity can support general fitness, joint movement, and well-being. Overexertion and high-impact exercise may damage already fragile muscles. A physiotherapist familiar with neuromuscular conditions can help identify a safe level and type of activity.

Can muscular dystrophy be cured with gene therapy?

Current gene-targeted treatments do not cure muscular dystrophy. Some may help selected

people with particular genetic variants, but they have limits and uncertainties. Research is continuing, and eligibility must be assessed by a specialist service.

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